GENETICS AND GENODERMATOSES

EPIDERMOLYTIC PALMOPLANTAR KERATODERMA (VöRNER TYPE)

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Background: The palmoplantar keratoderma (PPK) is a heterogeneous group of disorders of epidermal differentiation characterized by the presence of hyperkeratosis in palms and plants.
Can be autosomal dominant (AD), autosomal recessive (AR) and linked to the X chromosome. The differential diagnosis is based on the clinical signs, the association with other systemic or ectodermal disorders, the histology, the hereditary pattern and the underlying gene defect. Clinically, it is possible to identify three different patterns of hereditary PPK according to their distribution and extension: diffuse, focal and punctate.
The hereditary epidermolytic palmoplantar keratoderma was described by Vörner in 1901. It is AD, manifested during the first months of life, locating the mutation in the K9 and K14 keratin gene.
It is characterized by diffuse hyperkeratotic plaques with well-defined and erythematous borders, non-transgrediens, non-progrediens, fissures, hyperhidrosis and secondary dermatophytosis with absence of other associated ectodermal or organic alterations (simple PPK).

Observation: 65 year old woman. Congenital dermatosis that prevents fingerprinting.
Personal history: maternal grandfather and cousins with the same dermatosis.
Physical examination: diffuse hyperkeratotic plaques with well-defined limits, erythematous borders and fissures, which impedes hand mobility and erases the dermatoglyphs.
Histopathology: compact hyperkeratosis, acanthosis, papillomatosis and a pattern of epidermolysis that is manifested by the presence of keratinocytes of the spinous layers and granular with perinuclear vacuolization.
Laboratory; TG 240 mg / dl.
Diagnosis: Epidermolytic QPP of Vörner.
Due to hypertriglyceridemia, it was decided not to initiate systemic retinoids, but 40% Urea associated with topical 10% salicylic acid, with good response and decrease in keratoderma.

Key message: The PPK constitute a heterogeneous group of dermatoses. The importance of histology for diagnosis is highlighted.